GENETICS

Glossary and Indexing Instructions

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1968

acrocentric
syn. subterminal
type of chromosome in which the centromere is located near one end. At metaphase, it has the appearance of a "wishbone" (Illinois Med J 132:31, Jul 67)
Index CHROMOSOMES (68)

ALLELES (G1) 1968
syn. allelomorphs
alternative forms of the same gene, occupying the same place (=locus) on homologous chromosomes, governing the same characteristic of the phenotype (eye color, enzyme formation), and always segregating in meiosis (MeSH definition)

allelomorphs
see ALLELES

allocycly
the specific differential staining reactivity of heterochromatin which permits its cytological recognition and distinction from the remaining euchromatic portion of the chromosome (Illinois Med J 132:31, Jul 67)
Index CHROMOSOMES (IM) (68)
MITOSIS (NIM) (68)

anaphase
phase of cell division characterized by the movement of chromosomes from the metaphase plate towards the opposite poles (Illinois Med J 132:32, Jul 67)
Index MITOSIS (68)

ANEUPLOIDY (G1) 1968
syn. aneusomy
the chromosomal constitution of cells with an addition or subtraction of chromosomes to or from the normal set. Includes monosomy (symbol: 2N-1), TRISOMY (symbol: 2N+1), tetrasomy (symbol: 2N+2) (MeSH definition)

aneusomy
see ANEUPLOIDY

autosome
syn. heterosomes
a non-sex chromosome. Man has twenty-two pairs of autosomes and two sex chromosomes (XX or XY) (Illinois Med J 132:33, Jul 67)
Index CHROMOSOMES (68)

carrier, female
see female carrier

centric fusion
a special type of reciprocal translocation in which each of the involved chromosomes is broken very close to its centromere so that exchange of virtually entire, or even entire chromosome arms takes place ("whole-arm" translocation) (Illinois Med J 132:33, Jul 67)
Index CHROMOSOMES (IM) (68)
CROSSING OVER (NIM) (68)

centromere
syn. kinetochore
a non-staining apparently structureless constriction visible in chromosomes during prophase, metaphase and anaphase (Illinois Med J 132:34, Jul 67)
Index CHROMOSOMES (68)

chiasma
the point of contact between non-sister chromatids of paired homologous chromosomes, seen during the prophase of the first meiotic division. It is the cytologic expression of completed genetic crossing over, the latter providing for the exchange of genic material between two chromosomes before they separate (Illinois Med J 132:34, Jul 67)
Index CHROMOSOMES (IM) (68)
MEIOSIS (NIM) (68)
CROSSING OVER (NIM) (68)

chimera
an organism, usually a grafted plant, exhibiting a mixture of genetically different types lying adjacent to one another. This term has been applied to human subjects (twins) possessing more than one blood group: i.e., mixtures of A and O cells, who therefore exhibit erythrocyte mosaicism (Illinois Med J 132:34, Jul 67)
Index MOSAICISM (68)

CHROMATIDS (A11) 1968
the two microscopic subunits of chromosomes. Distinguish from DNA strand (MeSH definition; Illinois Med J 132:34, Jul 67)

chromatin
the substance in nuclei and chromosomes which stain intensely with basic dyes. It is composed of DNA combined with proteins. In the fixed intermitotic nucleus, it usually takes the form of an irregular network of long coiled threads. Larger and denser chromatin particles are known as chromocenters (Illinois Med J 132:34, Jul 67)
Index CHROMOSOMES (68)

chromatin-negative
refers to nuclei which lack Barr's sex-chromatin mass. This state is characteristic of the normal human male (Illinois Med J 132:34, Jul 67)
Index SEX CHROMATIN (68)

chromatin-positive
refers to nuclei containing the distinctive sex-chromatin body of Barr. It is present in the normal human female (Illinois Med J 132:34, Jul 67)
Index SEX CHROMATIN (68)

chromocenter
syn. sex-chromatin body of Barr
these are deeply staining clumps of chromatin material present in intermitotic nuclei. A chromocenter of characteristic size and shape lying in contact with the nuclear membrane is present in females of many species, including man(Illinois Med J 132:34, Jul 67)
Index SEX CHROMATIN (68)

CHROMOSOME ABERRATIONS (G1) 1968
deviations from the normal number or structure of chromosomes, not necessarily causing a disease. Structural aberrations include translocations, inversions, deletions, duplications, iso-chromosomes, and ring chromosomes (MeSH definition; Illinois Med J 132:35, Jul 67)

CHROMOSOMES (D2) 1968
one of the finite number of small bodies occurring in pairs into which the chromatin material of a cell nucleus resolves itself prior to cell division (Illinois Med J 132:34, Jul 67)

clone
a group of cells constituting the progeny of asexual reproduction from a single sexually-produced cell. The clone is of importance in mosaicism in which genotypically different ancestral cells give rise to genetically dissimilar stem-lines of daughter cells (Illinois Med J 132:35, Jul 67)
Index CLONE CELLS (68)

codominant (combinant)
refers to alleles whose effects are recognizable side by side, without one's being influenced by the other (Illinois Med J 132:35, Jul 67)
Index GENES, DOMINANT (68)

CONSANGUINITY (G1) 1968
blood relationship, this is, possession of common ancestors in the preceding few generations of the ascending line (Illinois Med J 132:35, Jul 67)

cross
see hybrid

CROSSING OVER (GENETICS) (G^1) 1968
an exchange of segments between chromatids of homologous chromosomes (MeSH definition; Illinois Med J 132:35, Jul 67)

deletion
a chromosomal aberration characterized by detachment and loss of a portion of a chromosome by breakage. In this manner, one or more genes may be removed from the organism (Illinois Med J 132:35, Jul 67)
Index CHROMOSOME ABERRATIONS (68)

deletions, chromosomes
see CHROMOSOME ABERRATIONS

DIPLOIDY (G1) 1968
the chromosomal constitution of somatic cells: two sets of chromosomes (symbol: 2N) (MeSH definition; Illinois Med J 132:35, Jul 67)

DNA (D10) 1968
essential component of the cell nucleus which carries the genetic information (Illinois Med J 132:35, Jul 67)

dominant
this classically refers to a gene which produces its effect despite the presence of an opposite or contrasting gene. Dominance and recessivity are complementary concepts. A gene is not dominant or recessive in itself but only in its behavior with respect to a certain allele. In human genetics, the homozygous state of "dominant" anomalies is usually unknown (Illinois Med J 132:35, Jul 67)
Index GENES, DOMINANT (68)

dominant, completely
applies to a gene whose heterozygous state cannot be distinguished from its homozygous state (Illinois Med J 132:35, Jul 67)
Index GENES, DOMINANT (68)

dominant, conditionally
applies to a gene only the heterozygous state of which is known, this state being markedly different, however, from the homozygous state of the other allele (Illinois Med J 132:35, Jul 67)
Index GENES, DOMINANT (68)

dominant, intermediately
applies to a gene which has a weaker effect in the heterzygous state than in the homozygous state (the expression has the same meaning as "semidominant, partially dominant, or incompletely dominant") (Illinois Med J 132:35, Jul 67)
Index GENES, DOMINANT (68)

dominant, irregularly
applies to a gene whose heterozygous state is not always recognizable. The greater the irregularity of the gene effect, the more questionable it is whether this is really due to a "dominant" gene and not rather to a more complicated genetic mechanism (Illinois Med J 132:35, Jul 67)
Index GENES, DOMINANT (68)

duplication
a type of chromosomal aberration characterized by the presence of an extra segment of chromosome which may exist as a separate fragment, or may be attached to one of the members of the ordinary chromosomal complement. This is to be distinguished from aneuploidy and polyploidy, in which variations in somatic chromosome numbers are irregular or exact multiples, respectively, of the basic chromosomal number (Illinois Med J 132:35, Jul 67)
Index CHROMOSOME ABERRATIONS (68)

equational division
the second meiotic division, essentially mitotic in type, characterized by the separation of sister chromatids. The latter are genetically identical since they are longitudinally-split reduplications of individual chromosomes (Illinois Med J 132:36, Jul 67)
Index MEIOSIS (68)

equatorial plate
syn. metaphase plate
a circular disc-like area situated at right angles to the spindle fibres of the dividing cell. On it are arranged the chromosomes preparatory to their separation (in meiosis I) or to the separation of their chromatids (potential daughter chromosomes) in mitosis, and in meiosis II. (Illinois Med J 132:36, Jul 67)
Index MITOSIS (68)

euchromatin
the chromatin material composing a chromosomal segment which exhibits no variation in staining intensity (iso-pycnosis) or in condensation at any time during the mitotic cycle. Most if not all genes directly responsible for hereditary transmission are believed to be situated in this type of chromatin. Contrasted with heterochromatin (Illinois Med J 132:36, Jul 67)
Index CHROMOSOMES (68)

euploidy
a state in which there is a balanced set of chromosomes. This includes variations in the somatic chromosomal number, characterized by the presence of the exact multiples of the basic number of the species. Accordingly, these may be the haploid (monoploid), diploid, triploid, tetraploid and higher multiple numbers. Complements higher than diploids are called polyploids (Illinois Med J 132:36, Jul 67)
Index GENES (68)

expressivity
the regularity in different individuals with which a gene produces a trait. Fluctuation in the degree of distinctness of a feature are explained by the "expressivity of the responsible gene." Low expressivity (low distinctness of a feature) passes continuously into "missing penetrance" (the feature is no longer recognizable) (Illinois Med J 132:36, Jul 67)
Index GENOTYPE (IM) (68)
GENES (IM) (68)

female carrier
a woman who is heterozygous for a recessive X-chromosomal gene. Half of the sons of a female carrier show the recessive X-chromosomal feature (Illinois Med J 132:160, Aug 67)
Index SEX CHROMOSOMES (IM) (68)
GENOTYPE (NIM) (68)

gamete
in bisexual organisms, the mature germ cell of either sex (spermatozoa or ova) containing the haploid ("n") number of chromosomes (Illinois Med J 132:160, Aug 67)
Index OVUM (IM) (68)
SPERMATOZOA (IM) (68)
if not specified GERM CELLS (IM) (68)

gene mutation
a mutation which involves only a single gene (Illinois Med J 132:160, Aug 67)
Index GENES (IM) (68)
MUTATION (IM) (68)

gene product
the specific protein or polypeptide molecules which depend on the presence of a gene are called primary gene products, frequently enzymes. Enzymes can synthesize most of the other molecules, such as complex polysaccharides, lipids, and even deoxyribonucleic acid. However, enzymes cannot synthesize proteins. For this, ribonucleic acid is required, which translates the information contained in the genes into the synthesis of the gene products (Illinois Med J 132:160, Aug 67)
Index GENES (IM) (68)
GENETICS, BIOCHEMICAL (NIM) (68)
or
specific enzyme or other chemical

GENES (G1) 1968
the basic ultramicroscopic intramolecular units of heredity, arranged linearly at definite points (loci) in a chromosome. They occur in pairs (allelic genes) situated at corresponding loci of a pair of homologous chromosomes and are capable of producing externally visible body traits composing the phenotype. Apparently, genes function by controlling enzymatic action (Illinois Med J 132:160, Aug 67)

genomic mutation
a mutation which alters the normal number of chromosomes of an individual. If the result of this is a whole multiple of the haploid set of the chromosomes, then one also speaks of "ploidic mutation" (Illinois Med J 132:160, Aug 67)
Index MUTATION (IM) (68)
CHROMOSOME ABERRATION (IM) (68)

GENOTYPE (G1) 1968
the genetic constitution of the individual; the characterization of the genes (MeSH definition; Illinois Med J 132:160, Aug 67)

gynandromorphism
the condition of sexual mosaicism in an organism of a bisexual species (Illinois Med J 132:160, Aug 67)
Index MOSAICISM (IM) (68)
SEX CHROMOSOMES (NIM) (68)

HAPLOIDY (G1) 1968
the chromosomal constitution of gametes (in humans spermatozoon and ovum): one set of chromosomes (symbol: N) (MeSH definition; Illinois Med J 132:164, Aug 67)

hemizygous
normal diploid cells, with their pairs of homologous chromosomes and genes, are either homozygous or heterozygous for each pair of genes, depending on whether the alleles of a pair are identical or contrasting. The hemizygous condition refers to the presence of an unpaired gene or chromosome. Hemizygosity, as an abnormality, exists in monosomic cells in which only one member of a pair of homologous chromosomes is present (Illinois Med J 132:161, Aug 67)
Index CHROMOSOME ABERRATIONS (IM) (68)
HAPLOIDY (NIM) (68)

hermaphrodite, pseudo-
see pseudo-hermaphrodite

hermaphrodite, true
an intersex that has gonadal tissue of both sexes present (Illinois Med J 132:161 Aug 67)
Index HERMAPHRODITISM (68)

HERMAPHRODITISM (C6,C7) 1968
the intersexual condition in which male and female genital structures exist in an individual of a bisexual species (Illinois Med J 132:161, Aug 67)

heterochromatin
the chromatin material occupying a chromosomal segment which exhibits variations in condensation and in staining intensity (heteropycnosis). Heterochromatin is generally believed to be lacking in genes directly concerned with hereditary transmission, although it probably exerts a modifying effect on near-by genes in the euchromatic zone (Illinois Med J 132:162, Aug 67)
Index CHROMOSOMES (68)

heterogametic
this refers to the XY or XO sex which produces two kinds of gamets (X, and Y or O) in equal numbers, whereas the XX series is said to be homogametic, since it produces only one kind of gamete (X) (Illinois Med J 132:162, Aug 67)
Index SEX CHROMOSOMES (68)

heteropycnosis
the phenomenon exhibited by the heterochromatic region of a chromosome, characterized by variations in the degree of condensation and in the intensity of staining (Illinois Med J 132:162, Aug 67)
Index CHROMATIN (68)

heteropycnosis, negative
occurs when the heterochromatic zone appears thinner and more faintly stained than the remainder of the chromosome (Illinois Med J 132:162, Aug 67)
Index CHROMATIN (68)

heteropycnosis, positive
occurs when the heterochromatic segment appears thicker and stains more intensely than the rest of the chromosome (Illinois Med J 132:162, Aug 67)
Index CHROMATIN (68)

heterosomes
see autosome

heterozygous
this refers to a zygote having a pair of dissimilar or contrasting alleles controlling a certain trait or characteristic (Illinois Med J 132:162, Aug 67)
Index HETEROZYGOTE (68)

holandric
this refers to genes situated in that portion of the Y-chromosome which does not pair with the homologous segment of the X-chromosome (Illinois Med J 132:162, Aug 67)
Index SEX CHROMOSOMES (68)

hologynic
inheritance from a woman to all female descendants. This occurs in Drosophila as a consequence of non-disjunction of the X-chromosome. In man, hologynic heredity is not known (Illinois Med J 132:162, Aug 67)
Index PHENOTYPE (68)

homogamy
occurs if chance does not prevail in matings within a population, but if like genes are joined with higher-than-chance frequency through mating (Illinois Med J 132:162, Aug 67)
Index BREEDING (68)

homozygous
this refers to a zygote having two members of a pair of genes alike for a certain trait or characteristic (Illinois Med J 132:162, Aug 67)
Index HOMOZYGOTE (68)

human genetics
see GENETICS, HUMAN

hybrid
syn. cross
the progeny of two parents who differ genetically in any way (Illinois Med J 132:162, Aug 67)
Index HYBRIDIZATION (68)

hyperploid
the aneuploid condition in which there is more than the normal diploid number of chromosomes, as exemplified by 2"n" plus 1, or 2"n" plus 2 in man with 47 or 48 chromosomes (Illinois Med J 132:162, Aug 67)
Index ANEUPLOIDY (68)

hypoploid
the aneuploid condition in which there is less than the normal diploid number of chromosomes, for example, the 2"n" minus 1 state in man with 45 chromosomes (Illinois Med J 132:162, Aug 67)
Index ANEUPLOIDY (68)

idiogram
a diagrammatic representation of the chromosome constitution of the cells of the individual. Customarily, the chromosomes are systematically arranged according to size and centromere position (Illinois Med J 132:163, Aug 67)
Index KARYOTYPING (68)

intermitotic
syn. interphase
the state of the resting nucleus, i.e., when the cell is not undergoing division. Chromosomes, as such, are not visible during this phase. Actually, the nucleus is not "resting" since biochemical activity is at its height at this time (Illinois Med J 132:163, Aug 67)
Index MITOSIS (68)

interphase
see intermitotic

intersex
(Illinois Med J 132:163, Aug 67)
Index HERMAPHRODITISM (68)

inversion
a structural chromosomal aberration characterized by the rearrangement (reversal) of a portion of the gene sequence. It occurs as a result of the breakage of a chromosome at two points followed by a reversal of the replaced fragment with respect to the rest of the chromosomes (Illinois Med J 132:163, Aug 67)
Index CHROMOSOME ABERRATIONS (68)

inversions, chromosome
see CHROMOSOME ABERRATIONS

isoalleles
alleles which, though so nearly identical that their effects cannot usually be distinguished, may have different modifying effects on genes inherited with them. This may influence the expressivity of the gene so that the disease varies slightly in different generations-in the father the abnormal gene will have been partnered by a different normal allele. The normal

alleles in the father and in his children are said to be isoalleles (Illinois Med J 132:163, Aug 67)
Index ALLELES (68)

isochromosome
an abnormal chromosome arising through the misdivision of the centromere. If the centromere misdivides, i.e., in a transverse rather than in a longitudinal plane, each of the two separated centromere fragments remains attached to the chromatids of one arm. Thus, the two new chromosomes (isochromosomes) differ from one another (Illinois Med J 132:164, Aug 67)
Index CHROMOSOME ABERRATIONS (68)

karyotype
a group of characteristics (number, size and form) used in identifying a particular chromosome constituion. For purposes of demonstration, a karyotype is a systematized array of the metaphase chromosomes of a single cell, typifying the chromosomes of an individual. It is customary to arrange the chromosomes in descending order of size, and according to the position of their centromeres (Illinois Med J 132:164, Aug 67)
Index KARYOTYPING (68)

kinetochore
see centromere

lethal factors
disorders of the genetic material .. which leads to the death of the zygote, or of the individual developing from it before attaining the age of reproduction. This can be a matter of lethal genes or deficiencies due to missing sections (Illinois Med J 132:164, Aug 67)
Index GENES, LETHAL (68)

lethal gene
a mutant gene which acts as a lethal factor (Illinois Med J 132:164, Aug 67)
Index GENES, LETHAL (68)

linkage
this term applies to two different genetic phenomena: linkage between a gene and a chromosome
Index GENES (IM) (68)
CHROMOSOMES (IM) (68)
and linkage between two genes (Illinois Med J 132:164, Aug 67)
Index GENES (68)

linkage
non-allelic genes which lie in the same pair of chromosomes are called linked. If they are in the same chromosome they are transmitted together to the descendents (exception: crossing over); if they lie in two different chromosomes of a homologous pair, a child obtains either one or the other gene (exception: crossing over has the result that a child may receive both genes). "Crossing over" is so frequent that no great practical significance is attached to linkage (Illinois Med J 132:164, Aug 67)
Index GENES (68)

median
syn. metagentric
this refers to the location of the centromere of a chromosome midway between the two extremities. The arms of the chromosomes are therefore of equal length (Illinois Med J 132:164, Aug 67)
Index CHROMOSOMES (68)

MEIOSIS (G1) 1968
a special method of cell division, occurring in maturation of the sex cells, by means of which each daughter nucleus receives half the number of chromosomes characteristic of the somatic cells of the species (MeSH definition; Illinois Med J 132:164, Aug 67)

metagentric
see median

metaphase
phase of cell division characterized by complete disappearance of the nuclear membrane and by the formation of the spindle. At this time, the chromosomes are maximally contracted and are arranged on the equatorial plate preparatory to separation. In mitosis, the metaphase chromosome has a single centromere which becomes functionally double only at anaphase. In the first meiotic division each bivalent has two centromeres. In the second meiotic division, metaphase is similiar to that of ordinary mitosis (Illinois Med J 132:165, Aug 67)
Index CELL DIVISION (68)

metaphase plate
see equatorial plate

MITOSIS (G1) 1968
indirect division of a cell, consisting of a complex of various processes, by means of which the two daughter nuclei normally receive identical complements of the number of chromosomes characteristic of the somatic cells of the species (MeSH definition)

monosomic
an aneuploid individual in whom there is a loss of one of the two members of a pair of homologous chromosomes. When it occurs spontaneously, this condition results from the meiotic non-disjunction, i.e., failure of the members of a pair to separate at anaphase (Illinois Med J 132:166, Aug 67)
Index ANEUPLOIDY (68)

mosaic
the presence of genetically dissimilar cells in adjacent tissue of an organism (Illinois Med J 132:166, Aug 67)
Index MOSAICISM (68)

MUTATION (G1) 1968
a change in a gene. The mutant gene is capable of transmission and thus of producing hereditary variations when it is present in germ cells. Mutation may also take place in somatic cells, in which case the mutant gene cannot be sexually transmitted. Mutant genes are usually recessive to the wild type (Illinois Med J 132:166, Aug 67)

mutation, gene
see gene mutation

2N
see DIPLOIDY

3N
see POLYPLOIDY

4N
see POLYPLOIDY

negative heteropycnosis
see heteropycnosis, negative

non-disjunction
a type of abnormal chromosomal behavior during nuclear division. It may occur in mitosis or in meiosis. As a result, one daughter cell receives both chromosomes while the other aquires none (Illinois Med J 132:166, Aug 67)
Index CHROMOSOMES (IM) (68)
CELL DIVISION (NIM) (68)

non-disjunction, primary
occurs in one or both parents, with the production of gametes containing an abnormal chromosomal constitution (Illinois Med J 132:166, Aug 67)
Index CHROMOSOMES (IM) (68)
CELL DIVISION (NIM) (68)

non-disjunction, secondary
refers to the occurrence of non-disjunction during meiosis in an individual who is abnormal from the beginning, being a product of the union of gametes, one of which was non-disjunctional (Illinois Med J 132:166, Aug 67)
Index CHROMOSOMES (IM) (68)
CELL DIVISION (NIM) (68)

penetrance
the regularity with which a gene produces its effect. Penetrance is "complete" if a dominant gene produces its effect in each person carrying it, or if a recessive gene causes its effect in each individual homozygous for it. It is "reduced" if some but not all persons exhibit the trait even though the gene is present in the required heterozygous or homozygous stage (Illinois Med J 132:166, Aug 67)
Index GENES (68)

phene, phenic
the features which are assigned to certain genes or configuration of genes are called phenes. The expression is not equivalent to phenotype (Illinois Med J 132:166, Aug 67)
Index PHENOTYPE (68)

phenocopy
a reproduction or copy of a gene-producing phenotype which is visibly indistinguishable from it. It occurs in the offspring of pure normal individuals as a non-hereditary modification produced by certain experiemental conditions (Illinois Med J 132:166, Aug 67)
Index PHENOTYPE (68)

phenogenetics
the science which attempts to explain the chain of causality between genotype and phenotype. Phenogenetics is more a part of developmental physiology than of genetics (Illinois Med J 132:168, Aug 67)
Index GENETICS (68)

PHENOTYPE (G1) 1968
the outward appearance of the individual; the product of interactions between genes and between the genotype and the environment (MeSH definition; Illinois Med J 132:168, Aug 67)

pleiotropism
a gene which determines several characteristics is called pleiotropic or polyphenic. A gene might have a major effect, such as that controlling the production of a blood group antigen and also play a minor role in predisposing to duodenal ulcer (Illinois Med J 132:168, Aug 67)
Index GENES (68)

ploidic mutation
see genomic mutation

point mutation
a mutation which is not accompanied by a morphologically demonstrable change in the chromosome. In practice, it may be regarded as equivalent to a gene mutation (Illinois Med J 132:168, Aug 67)
Index MUTATION (68)

polygenic
a feature, usually a quantitatively variable character, is polygenic if it is dependent on numerous genes. Most quantitatively variable physiological characteristics have polygenic inheritance. The involved genes are also designated as polygenes (Illinois Med J 132:168, Aug 67)
Index PHENOTYPE (IM) (68)
GENES (NIM) (68)

polymerism (genetics)
polygenic effect in which the participating genes are of equally strong effectiveness (Illinois Med J 132:168, Aug 67)
Index PHENOTYPE (IM) (68)
GENES (NIM) (68)

polymorphism
the simultaneous occurrence of two or more different genotypes in the population whose ratio is kept in equilibrium by means of selection (Illinois Med J 132:168, Aug 67)
Index GENOTYPE (68)

polyphenin
(Illinois Med J 132:169, Aug 67)
Index GENES (68)

POLYPLOIDY (G1) 1968
the chromosomal constitution of a cell containing multiples of the normal chromosomal set; includes triploidy (symbol: 3N), tetraploidy (symbol: 4N) etc. (MeSH definition; Illinois Med J 132:168, Aug 67)

polysomic
an individual, an aneuploid in whom one or more chromosomes are reduplicated and are found represented three or four times. The abnormal number of chromosomes results from meiotic non-disjunction. The resulting zygote has a 2"n" plus 1 chromosomal constitution. Such a polysomic organism is also designated trisomic for the non-disjunctional chromosome (Illinois Med J 132:168, Aug 67)
Index ANEUPLOIDY (68)

population genetics
see GENETICS, POPULATION

positive heteropycnosis
see heteropycnosis, positive

primary non-disjunction
see non-disjunction, primary

proband
syn. propositus
the index patient, an individual with an abnormality whose relatives are studied in an effort to ascertain the hereditary or genetic aspects of the trait (Illinois Med J 132:168, Aug 67)
Index GENOTYPE (68)

prophase
the first phase of cell division characterized by the transformation of the irregular network of chromatin material of the interphase nucleus into spirally coiled threads, which gradually become doubled, shortened, and thickened to form individual chromosomes (Illinois Med J 132:168, Aug 67)
Index CELL DIVISION (68)

propositus
see proband

pseudo-hermaphrodite
an intersex that has the gonads containing either testicular or ovarian tissue respectively, but not both (Illinois Med J 132:161, Aug 67)
Index HERMAPHRODITISM (68)

recessive
this refers to a gene which fails to produce its effect in the presence of the opposite or contrasting gene. A recessive gene produces its effect only when both members of a pair are recessive, i.e., when the individual is homozygous for this particular gene, having received it from both parents (Illinois Med J 132:168, Aug 67)
Index GENES, RECESSIVE (68)

recombination
the union of two members of a pair of genes for a given character. This occurs at fertilization, each gamete contributing its single member of a pair of genes (Illinois Med J 132:168, Aug 67)
Index RECOMBINATION, GENETIC (68)

reductional division
the first meiotic division, characterized by the mere separation of members of homologous pairs of chromosomes, resulting in a reduction in the chromosome number from the somatic or diploid number to the haploid number (Illinois Med J 132:169, Aug 67)
Index MEIOSIS (68)

ring chromosome
see CHROMOSOME ABERRATIONS

satellite
a small rounded body attached to the end of certain chromosomes by a delicate thread of chromatin. Satellites and their filaments are constant in their size and form for each particular chromosome. Therefore, they provide landmarks for the identification of these chromosomes (Illinois Med J 132:169, Aug 67)
Index CHROMOSOMES (68)

secondary non-disjunction
see non-disjunction, secondary

segregation
the process in sexual organisms, constituting the Mendelian first law of inheritance, by which only one of the two genes of a pair gets into each mature germ cell. Then, when gametes unite at fertilization, two genes for each trait or character are brought together in the new individual. Segregation takes place during the reductional stage of meiosis (Illinois Med J 132:169, Aug 67)
Index MEIOSIS (68)

sex-chromatin body of Barr
see chromocenter

SEX CHROMOSOMES (All) 1968
a pair of chromosomes determining the sex of the individual. One sex has 2 equal sex chromosomes (XX in the human and Drosophila female), while the other has different sex chromosomes (XY in the human and Drosophila male). Distinguish from SEX CHROMATIN (MeSH definition; Illinois Med J 132:169, Aug 67)

sex-influenced
this refers to genes which are dominant in one sex and recessive in the other, such as those for baldness, and for one form of white forelock (Illinois Med J 132:169, Aug 67)
Index SEX CHROMOSOMES (IM) (68)
GENES (NIM) (68)

sex-limited
this refers to genes, the penetrance of which is completely reduced in one sex, i.e., they are capable of producing traits in one sex but not in the other (Illinois Med J 132:169, Aug 67)
Index SEX CHROMOSOMES (IM) (68)
GENES (NIM) (68)

sex-linked
this refers to genes located in a sex chromosome, X or Y. The best known sex-linked genes in man are those for red-green color blindness, and hemophila (Illinois Med J 132:169, Aug 67)
Index SEX CHROMOSOMES (IM) (68)
GENES (NIM) (68)

somatic mutation
a mutation which involves a somatic cell and its daughter cells, that is, only a part of the organism (Illinois Med J 132:276, Sept 67)
Index MUTATION (68)

specificity
a gene which can have quantitatively differentiated effects in different individuals has a low "specificity." Since this practically never occurs, the expression can be dispensed with. The high degree of specificity of gene effects is a matter of general experience in human genetics (Illinois Med J 132:276, Sep 67)
Index GENES (68)

spindle
a fusiform bundle of delicate filaments appearing at the beginning of the metaphase stage of cell division (Illinois Med J 132:276, Sep 67)
Index CELL DIVISION (68)

submedian
this refers to the location of the centromere of a chromosome at a point elsewhere than at the center, or near the end (Illinois Med J 132:276, Sep 67)
Index CHROMOSOMES (68)

subterminal
see acrocentric

supernumerary chromosome
one or more extra chromosomes found inconstantly in wild populations of certain species of animals. They are not homologous to members of the regular set of chromosomes and they apparently exert little influence on the phenotypic effect (Illinois Med J 132:276, Sep 67)
Index CHROMOSOMES (68)

synapsis
the period in meiosis in which the two members of every pair of homologous chromosomes are in very precise and intimate apposition (Illinois Med J 132:276, Sep 67)
Index MEIOSIS (68)

syngamy
in sexual reproduction, the union of germ cells (gametes) at fertilization to produce a new individual, a zygote (Illinois Med J 132:276, Sep 67)
Index GERM CELLS (68)

telophase
the final phase of cell division during which the chromosomes regroup into a nuclear structure and are gradually transformed into long thread-like fibers enclosed by a nuclear membrane (Illinois Med J 132:276, Sep 67)
Index CELL DIVISION (68)

tetraploid
an organism containing four complete sets of each kind of chromosome characteristic of the species. Thus, the tetraploid number in man is 92 (Illinois Med J 132:276, Sep 67)
Index POLYPLOIDY (68)

translocation
a type of aberration characterized by fragmentation of a chromosome and transfer of the broken-off portion to another chromosome, often of a different pair (Illinois Med J 132:277, Sep 67)
Index CHROMOSOME ABERRATIONS (68)

translocations, chromosome
see CHROMOSOME ABERRATIONS

triploid
an organism containing three complete sets of each kind of chromosome characteristic of the species. Therefore, the triploid number in man is 69 (Illinois Med J 132:278, Sep 67)
Index POLYPLOIDY (68)

trisomic
an aneuploid (hyperploid) individual in whom a chromosome is present in triplicate, rather than in the normal double or disomic number, or condition. (Illinois Med J 132:276, Sep 67)
Index TRISOMY (68)

TRISOMY (C16) 1966
numerous congenital anomalies that are the result of the presence of a third chromosome in one of the pairs of the autosomes (trisomy), so that the total number of chromosomes in the karyotype of the affected individual is likewise 47. The three autosomal trisomes known at the present time are: trisomy 21 or mongolism, with a small acrocentric chromosome in 21; trisomy 13-15 or D_1 syndrome with a large acrocentric chromosome in 13-15: and trisomy 17-18 or E syndrome with a medium sized acrocentric chromosome in 17-18 (MeSH definition)
Index all trisomies
TRISOMY (IM) (68)
CHROMOSOMES, HUMAN (IM) (68)
and
specific chromosome number

trisomy 13-15
Index TRISOMY (IM) (68)
CHROMOSOMES, HUMAN, 13-15 (IM) (68)

trisomy 17-18
Index TRISOMY (IM) (68)
CHROMOSOMES, HUMAN, 17-18 (IM) (68)

trisomy 21
Index MONGOLISM (68)

true hermaphrodite
see hermaphrodite, true

zygote
the cell resulting from the fusion of two gametes; the fertilized ovum (Dorland)
Index OVUM (IM) (68)
FERTILIZATION (NIM) (68)